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Abstract

Throughout our history, science was always on the front lines for discovery and exploration. Science is used as an investigative tool by the human race to figure out all the mysteries of the universe. The discovery of DNA was tremendous, providing each human being with their own unique genetic identity - no longer would an individual be genetically confused with another. DNA fingerprinting, in particular, has changed the world. In the 1980's the legal system began using DNA fingerprinting to help establish the guilt of an indicted criminal. DNA (besides for fingerprints) is the only way to confirm scientifically if the individual was at the scene of the crime. Over the years, many methods for forensic DNA testing have emerged. Polymerase chain reaction is a method used to amplify the smallest amounts of DNA, creating thousands of copies which can be analyzed. Restriction fragment length polymorphism looks for variations in homologous DNA. Short tandem repeat technology looks for repeated sequences in the bases of the DNA sample. Mitochondrial DNA analysis tests the mitochondrial genome which is highly polymorphic between individuals. Finally, Y-Chromosome analysis is used for males, and usually accompanies PCR or RFLP. DNA is now commonly used in criminal investigations and is often the most substantial piece of evidence. In recent years, DNA fingerprinting is also being used for exonerations. People who have been languishing in prison for years for crimes they did not commit are being released due to the breakthrough of forensic DNA and DNA fingerprinting establishing their innocence.

"The blood or semen that (the perpetrator of the crime) deposits or collects- all these and more bare mute witness against him. This is evidence that does not forget. Physical evidence cannot be wrong; it cannot perjure itself; it cannot be wholly absent. Only human failure to find, study and understand it can diminish its value."

(Paul Kirk, Crime Investigation, 1953)

Introduction: Forensic DNA

Forensic DNA is an identification system that allows DNA typing to be performed on an extremely minute amount of organic human matter. The DNA can be extracted from bloodstains, hair, saliva, debris from fingernail, teeth, dandruff, epidermal cells, fingerprints, personal items, and more. Forensic analysis of DNA is a commonly used - though relatively recent method - of helping to identify the victim or perpetrator in criminal investigations. In modern crime investigation, once a crime is committed, forensic protocols swing into action, with police and specialized teams that analyze and comb through all available evidence. Upon discovering possible DNA evidence, it is collected with the greatest importance given to keeping it sterile and untainted. Once all the DNA has been collected, it is brought to a lab that will determine which method of DNA typing will be performed, based on the quantity of DNA collected.

DNA evidence, as it is now used, is a very powerful investigative tool when at a crime scene. DNA is strong, concrete evidence which can help link a suspect to the crime, or, in the alternative, prove that a certain individual was not present at the scene of the crime. Because of this, the combination of forensic science utilizing the properties of DNA is taking up an ever-increasing role in the investigation of crimes. DNA is collected routinely, and is many times the key investigative evidence sought after and used by the authorities. Although DNA testing can take anywhere from one week to 3 month to obtain results, its high rate of accuracy is well worth the wait. The process of comparing DNA linked to a crime is simple; one sample is taken from the suspect, and one sample is taken from the crime scene. The DNA from both samples is studied and compared, and if the DNA matches, then there is near complete certainty that the tested individual was present at the crime scene.

While it may seem to be a boon for prosecutors looking to put criminals behind bars, DNA evidence is also being used with great success to clear individuals of a previous convicted guilt. Many cases that were brought to trial when DNA evidence testing was not a viable criminal justice method have been retried based on evidence obtained through DNA testing. Evidence of DNA at the crime scene, however, is not absolute proof. While DNA testing establishes with almost complete certainty the presence of an individual's DNA to the crime scene, there still remains the possibility for sample error. For example, a person's DNA could have been at the crime scene before the crime had been committed, or an accused person can have DNA nearly identical to a relative, such that the sequencing may be an almost exact match. Due to this uncertainty, there is significant debate over the level of weight DNA evidence should be granted in courtroom proceedings.

What is DNA?

Deoxyribonucleic acid, or DNA, is a very powerful molecule present in each human being. Furthermore, DNA is unique to each individual, so that no two people (besides identical twins) share the same precise DNA combination. The rainbow of different human attributes, from physical to mental traits, is due to the unique DNA we all carry. The structure of DNA was first discovered by Watson and Crick in 1953. They found that DNA had two parallel strands that took the shape of a double helix. The parallel strands, each made up of four nucleotides (adenine, thymine, guanine and cytosine) chained together in a specific sequence. The backbone of DNA is composed of alternating sugar and phosphate residues (figure 1). The sugar in the

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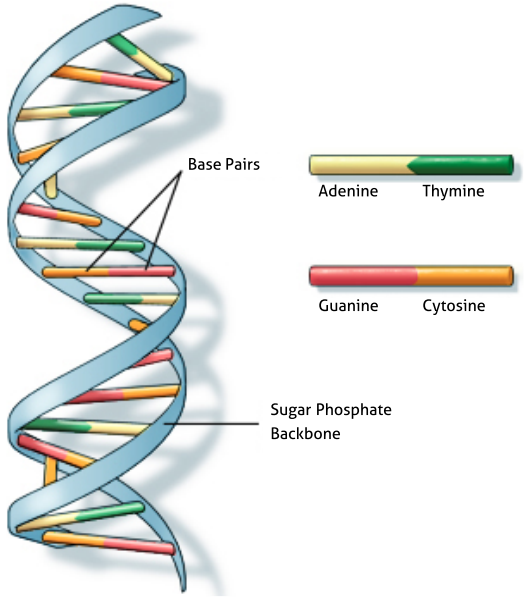

Figure 1: DNA Helix
Source: U.S. National Library of Medicine

backbone is a 5 carbonsugar (pentose) which is called 2-deoxyribose. Phosphate groups are what hold the sugar molecules together, and provide the strand of DNA with a direction. DNA is anti-parallel; the directionofone strandof nucleotides is the opposite direction of their bonded strand. There is a 5 prime end which has a phosphate group and a 3 prime end which has a hydroxyl group. So while one strand is going from 5 prime to 3 prime, the second strand is going from 3 prime to 5 prime. DNA sequence is virtually the same in every single cell within a person's body. However, while all humans share 99% of their DNA with everyone else it is a mere 1% difference that differentiates one person from the other. There is only 1 difference in every 1,200 to 1,500 nucleotides. But that one difference is found in sufficient quantity to allow for tremendous variety.

The difference from one individual's DNA to another's is in their genome, which is the complete set of DNA, including all of its genes. Genome variations are differences in a person's DNA sequence. Generally, most genome differences are simple, only involving variations in a few bases. Each of these sequence difference is called a DNA polymorphism. In addition to the genome variation of different bases, there is another DNA polymorphism involving the number of repetitions of a particular sequence of nucleotides.

DNA within the nucleus of each cell encodes the genetic instructions for all development and functioning of each being. It is the building block for each individual's genetic make up. DNA is one of the major macromolecules essential for all known forms of life. Genetic information is encoded as a sequence of the four nucleotides. DNA is different from one person to the next, but within each person their DNA is the same in every cell within their body.

Collection and Extraction of DNA

The process of DNA analysis begins with the extraction of the DNA. The DNA must be separated from from all other cell components. "There are various possible DNA extraction methods and when dealing with crime scene samples the type of evidence and the amount of DNA it contains will help determine the extraction method used. Common forensic DNA extraction methods include the use of chelex beads" (Kobilinsky, 2011). Chelex beads are ion- exchange resins that protect DNA by binding to magnesium ions. This process inhibits magnesium from destroying DNA. DNA is released from the cell, after the cells have been broken open by boiling the cellular material in the presence of chelex beads. After this process, the cells are placed in a centrifuge and all cellular material - including the chelex beads - fall to the bottom of the tube, while the liquid in the tube contains the extracted DNA. The liquid is usually transferred to a new tube, where it is frozen at -20 or -80 celsius until it is used for analysis. Another method for DNA extraction is Organic Extraction. This involves adding chemicals to the DNA sample. "First sodium dodecylsulfate (SDS) and proteinase K are added to break open the cell wall, and to break down the proteins that protect the DNA molecules while they are in chromosomes. Next a phenol/chloroform mixture is added to separate the proteins from the DNA. The DNA is more soluble in the aqueous portion of the organic-aqueous mixture. When centrifuged, the unwanted proteins and cellular debris are separated away from the aqueous phase, and double stranded DNA molecules can be cleanly transferred for analysis" (Butler, 2005).

Before being analyzed in the lab, the amount of human DNA must be measured. This is due to that fact that all kinds of DNA are collected from a crime scene, not only human DNA. So the DNA Advisory Board standards require human specific DNA quantitation. The most common process is called the "slot-blot" procedure. This test is specific for human DNA. On a nylon membrane with addition of a human specific probe, genomic DNA is captured. It is a measurement of the comparison between the unknown samples to a set of standards.

Methods for Forensic DNA Profiling

Polymerase Chain Reaction

There are many methods of forensic DNA testing used to analyze the evidence. The one most commonly used today is Polymerase Chain Reaction (PCR). It is the most commonly used method due to its practicality - only a small sample is required, and it can be done on samples that have not been recently collected. In fact, PCR can be performed on old samples of DNA many years later. It was developed by Kary Mullis in 1983. In the PCR test, biochemical technology is used to amplify a small sample of nuclear DNA to millions of copies of a particular DNA sequence. There are several steps in this procedure. First the DNA has to be denatured. Denaturation separates the complementary strands of DNA held together in the duplex by hydrogen bonds. Thus, samples are heated to 94°-96° Celsius for one to two minutes until the DNA is separated into single

strands. This works because the strands are bonded together with a weak hydrogen bond, as the sugar and phosphate backbone is bonded together with a strong covalent bond. Next, in the annealing step, the temperature is lowered to 50º-65º Celsius, and primers bind to the DNA. "A primer is a single stranded sequence of nucleotides known as an oligonucleotide. Each primer is complementary to one of the original DNA strands to either the left 5 prime side, or right 3 prime side of the sequence of interest." (Schochetman, Ou, Jones. 1988) The primer binds to the primer template and acts as a starting point for DNA formation. Two primes are involved in PCR, one for each strand. Next is the extension step where new DNA strand is synthesized complementary to the DNA template. At the end of this cycle, there are two new DNA strands identical to the original target sequence. These DNA strands are called Amplicon. The extension step can vary in time and cycles. It depends on DNA polymerase used, and the length of the DNA fragment to be amplified. After a few cycles of this, the target sequence of the original DNA strand is amplified.

Restriction Fragment Length Polymorphism

The second method is Restriction Fragment Length Polymorphism (RFLP). It was first discovered in the 1980's by Alec Jeffreys. This was the first method in testing forensic DNA. Jefferys was working on DNA profiling (DNA fingerprinting). He used the difference in length of nuclear DNA regions created by variations of numbers in repeated sequence to distinguish between individuals. Restriction enzymes recognize specific sequences of nucleotides in DNA called restriction endonuclease recognition sites. "The enzymes that are commonly used for restriction fragment length polymorphism analysis require 4-6 base pair recognition sequences. Cleavage frequency can be estimated by making the assumption that each of the different nucleotides occur randomly and in equal amounts for a given DNA sequence" (Bernatzky, 1988). The enzyme cuts the DNA in a process known as restriction digest. DNA's restriction sites, and distances between the sites differ from one person to the other. These differences are called restriction fragment length polymorphism. By using a restriction enzyme to cut a DNA sample, different lengths are obtained. The resulting pieces of DNA are passed through Agarose gel electrophoresis, which sorts out a pattern of bands by length that is unique for the particular DNA being analyzed. These repeated regions of DNA are called Variable Number Tandem Repeats. The fragments of DNA are transferred to a sheet of nitrocellulose which is exposed to a radioactive probe. After, a photographic film is laid on top of the sheet to expose an image corresponding to the DNA fragments. RFLP occurs when the detected length varies between individuals." RFLP analysis has been used for a variety of purposes. Since restriction sites are actual samples of nucleotide sequence the variation for the presence of sites has been used to estimate genetic divergence of individuals" (Bernatzky, 1988). Each fragment length is considered an allele, and has genetic property to it. This method is not used very frequently for forensic DNA because a fairly large sample is needed; a sample of 100,000 cells or more. Another downside to RFLP is that the DNA sample needs to be "fresh" from the crime scene, and as a result, this method cannot be performed on old DNA samples. Although PCR is used more often, RFLP is considered a more accurate test.

Short Tandem Repeat Technology

The third method is Short Tandem Repeat Technology (STR), which is also referred to as Microsatellites, or Simple Sequence Repeats (SSR). It was introduced in the late 1990's. It's used for the analysis of specific regions found in nuclear DNA. STR's is a type of polymorphism where short sequences of tetra or penta nucleotide repeats of DNA are repeated and the repeated sequences are adjacent to each-other. The pattern can range from 2 to 10 base pairs, and is typically in the non-coding intron region, making the DNA unimportant. STR's are not considered so important because they do not code for a protein. By looking at many STR loci and counting how many specific repeats there are, it is possible to create a unique genetic profile for individuals. Once a STR has been found, the PCR process is often used to amplify that specific sequence. Once these sequences have been amplified, they are put through gel electrophoresis. After, the DNA is placed under a fluorescent dye to be visualized.

Mitochondrial DNA Analysis

Another method for testing forensic DNA is called Mitochondrial DNA Analysis. It is used when the DNA evidence is not suitable for PCR, STR, or RFLP. This mitochondrial DNA is present in the mitochondria of every human cell. It is very different than nuclear DNA. Mitochondrial DNA is useful for forensic purposes because it has two properties. Firstly, the mitochondrial genome is highly polymorphic, which is very helpful when is comes to human identification. Secondly, It's genes exist in a high concentration even though mtDNA only makes up for only 1% of the DNA within a cell. This is very useful for old or degraded DNA that needs to be tested and lacks nuclear DNA. In addition, mtDNA is strictly inherited from the maternal side. Therefore all siblings have the exact same mtDNA in the absence of a mutation. It comes in handy in a missing persons investigation, but it has a down side to it. There is no differentiation between mother and all her offspring.

Y-Chromosome Analysis

The last method for discussion is called Y-Chromosome Analysis. This method usually performed as an adjunct to one of the other discussed tests. In particular, the Y-Chromosome analysis is useful in cases involving sexual or paternal allegations. The Y-Chromosome is passed directly from father to son, which can provide determinative biological evidence involving multiple male contributors. Although this may be somewhat useful in a crime case, it usually used when trying to find familial relationships.

DNA Under the Law of Scientific Evidence

With the new technologies for forensic DNA, the courts have applied many standards to make sure the reliability of the evidence is true. There are two types of regulations. The Frye

rule requires all scientific evidence to be "generally accepted" by the scientific community before being admitted into the courtroom. The second standard is the Federal Rules of Evidence (FRE), which the Supreme Court ruled superseded the Frye standard. The FRE requires the scientific evidence to be helpful and relevant.

Ever since DNA has been admitted into the courtrooms, there have been more guilty verdicts. DNA evidence is extremely helpful and useful in solving a crime and finding the guilty party, but it not completely sufficient yet. Forensic DNA evidence is nearly, but not 100% accurate. Yet as the years pass, new technology is being introduced to achieve the 100% standard for scientific evidence. Eventually the technology will compel complete acceptance of DNA evidence.

Case Study: The O.J. Simpson Murder Trial

On June, 13 1994 Nicole Brown Simpson and Ronald Goldman were found dead outside Brown's house. All the evidence collected from the scene led the police to suspect that O.J. Simpson was the person who committed the murder. At this point, forensic DNA was relatively new, and not always accepted or believed as concrete evidence. "Simpson's lawyers are expected to mount a vigorous assault on the validity of forensic DNA evidence in an effort to convince Judge Lance to keep it out of court'" (Norwak, 1994). There was a great amount of forensic evidence that proved O.J. Simpson was indeed the murderer.

There was a great amount of strength and weakness of the DNA evidence against O.J. Simpson. The prosecution found that O.J. cut his hand during the murder, and left a trail of blood from the murder, to his car and into his house. There was also Nicole's blood found on bottom of his sock. There was also a glove found in Simpson's house that was covered in Nicole and O.J.'s blood. During the trial, Barry Scheck, a lawyer who specializes in forensic DNA, spent eight full days questioning the forensic evidence that was collected and tested. During this cross-examination, several aspects were brought to light about the collection of the DNA evidence that created doubt as to the accuracy of the sample obtained. The defense was able to debunk all of this evidence. They stated that the glove which had Nicole and O.J.'s DNA on it was contaminated at the LAPD laboratory. LAPD lab criminalist Collin Yamauchi admitted that the glove was indeed contaminated, and that he accidentally spilled a vial of O.J.'s blood on the glove. "The criminalists were poorly trained with respect to sample handling, were not following a written protocol, did not understand the purpose and importance of precautionary measures, such as changing gloves and made serious errors when attempting to demonstrate proper sample collection and handling techniques" (Thompson, 1996). The defense alleged that the DNA evidence was indeed tampered with, or not processed correctly. It was found that Andrea Mazzola had collected a blood DNA sample from O.J. Simpson, but had let that sample sit in her lab coat pocket the entire day before returning it to the lab for testing. Barry Schenck was able to convince the jury that the forensic evidence was not handled correctly, and that there was a reasonable doubt that it could be relied upon in proving Simpson's guilt.

The O.J. Simpson trial was one of the first trials that concentrated much of it's efforts in the areas of forensic fingerprinting. While ultimately the DNA evidence was not accepted by the jury, the publicity of the trial created a far greater awareness of the methodology and its great power as evidence.

Project Innocence: Exoneration

"In New Jersey, March of 1988, Byron Halsey was convicted for the brutal rape and murder of a seven year old girl and an eight year old boy. The evidence used to convict Halsey was his supposed confession, which he gave after over thirty hours of interrogation and sleep deprivation. Halsey had to "guess several times" before he could correctly describe to police how the crime occurred and other key factors...They jury convicted him using that evidence. After nineteen long years in prison, newly analyzed DNA test results proved Halsey's innocence and implicated the actual killer" (Sophia Chang, 2009).

Aside for the conviction purposes, recently DNA has been widely used for exoneration purposes. It is true that DNA has so much power that it can send someone to prison. But it also possesses the same amount of power in setting a man who was wrongly convicted free.

The innocence project was founded in 1992 by Barry Schenck and Peter Neufeld at the Cardozo School of Law at Yeshiva University. They came up with this idea to assist people who can be proven innocent through proper DNA testing. Before they take on a case, they do extensive screening to see if there is proper DNA to be tested. "DNA testing has opened a window into wrongful convictions so that we may study the causes and propose remedies that may minimize the chances that more innocent people are convicted" (The Innocence Project). To date there have been nearly 300 prisoners in the United States that have been exonerated because of DNA testing.

Conclusion

Science is in constant state of evolution. The first big break in forensic evidence was fingerprinting, which was discovered over 100 years ago. The next big discovery for forensic evidence was DNA fingerprinting. Since the development of forensic DNA testing in the early 1980's it's sophistication and accuracy has continuously improved, so that it is now considered a fundamental part of any investigation. The methods for analyzing DNA evidence are quite varied, with unique advantages and disadvantages to each. Yet, they are all really about one thing-the cataloging in DNA of the unique attributes of every person. By utilizing this method, there is a far greater likelihood of the investigations leading to the actual perpetrator. One needs to look no further than the many exonerations due to the Innocence Project to see how relatively primitive previous investigative methods are in comparison to DNA forensics. The careful study of the mechanics of DNA and

its attendant forensic methods will likely yield every greater scientific results in the future.

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